Hematologically Important Mutations: Glucose-6-phosphate Dehydrogenase

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A total of 97 mutations or combination of mutations of the X-linked gene for glucose-6-phosphate dehydrogenase are now known to us. Previous tabulations, providing somewhat more detailed information about these mutations (1,2) and comprehensive reviews of the deficiency (2,3) may be found in the cited sources and in OMIM on the World Wide Web (http://www3.ncbi.nlm.nih.gov/htbin-post/Omim/dispmim?305900)

The cDNA numbers shown in the table are based on the Genbank sequence with the

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Editor's note: *The pace at which mutations are being found has become so rapid that the publication of up-to-date tabulations has become impossible using conventional means. For example, the last comprehensive publication of glucose-6-phosphate dehydrogenase variants (reference 2 in the present paper) was submitted for publication on June 1994 and published in December of 1994. Since its publication 30 additional variants have been discovered and many that were unpublished in the earlier tabulation have now appeared in print. Since papers are published in this journal within less than two weeks of receipt, reprints are available in 2 weeks, and the printed version is on library shelves within an average of 3 months, it provides an ideal medium for publication in such a rapidly moving field.*

Updated registries of some mutations are beginning to appear on the internet, and eventually this may be the common means for providing access to mutations. In the meanwhile, the publication of up-to-date, authoritative lists of mutations in Blood Cells, Molecules and Diseases *can fill the need. The commentaries that we encourage and publish provide an opportunity for these tabulations to be updated and, where needed, corrected by the readers. These corrections will also appear on the internet and in the journal and become a part of the permanent record documenting the mutations covered. We encourage colleagues who are maintaining mutation databases to submit them to us for rapid publication. In this way we hope to provide those interested in the genetics of hematologic disease with a readily available and very up-to-date source of information concerning what mutations have been found and where more information can be obtained about them.*

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accession number X03674 and the genomic numbers on the sequence with accession number X55448. In each case the A of the ATG start codon is numbered +1. Note that this numbering is different from that used in the GenBank files. It is necessary to subtract 470 from the GenBank cDNA sequence and 3350 from the GenBank genomic sequence to standardize these sequences to begin at the start codon.

Table: G6PD variants that have been characterized at the DNA level

Variant Name*	cDNA Nucleotide Substitution	Genomic Nucleotide Substitution	Amino Acid Substitution	References
Gaohe Gaozhou	95 A→G	95G	32 His→Arg	(4)
"Honiara"	99 A→G 1360 C→T	99G 13763T	33 Ile→Met	(5)
"Sunderland"	105-107 del	105-107 del	105-107 del	(6)
"Orissa"	131 C→G	9988G	44 Ala→Gly	(7)
"Aures"	143 T→C	10000C	48 Ile→Thr	(8)
"Kozukata"	159 G→C	10111C	53 Trp→Cys	(9)
"Kamogawa"	169 C→T	10121T	57 Arg→Trp	(9)
Metaponto	172 G→A	10124A	58 Asp→Asn	(10)
A- Distrito Federal "Matera" Castilla Alabama Betica Tepic Ferrara Laghout Kabyle	 202 G→A 376 A→G	 10154A 10877G	 68 Val→Met 126 Asn→Asp	(11) (12) (10) (12) (13) (14) (12) (15) (16) (16)
Namoru	208 T→C	10160C	70 Tyr→His	(17)
Murcia	209 A→G	10161G	70 Tyr→Cys	(18)
Swansea	224 T→C	10176C	75 Leu→Pro	(19)
Ube Konan	241 C→T	10193T	81 Arg→Cys	(20)
"Lagosanto"	242 G→A	10194A	81 Arg→His	(21)
Urayasu	281-283 AGA del	10782-10784	94 Lys del	(22)

"Vancouver "	317 C→G 544 C→T 592 C→T	10818G	106 Ser→Cys 182 Arg→Trp 198 Arg→Cys	(23)
São Borja	337 G→A	10838A	113 Asp→Asn	(24)
A	376 A→G	10877G	126 Asn→Asp	(25)
Vanua Lava	383 T→C	10884C	128 Leu→Pro	(17)
"Chinese-4"	392 G→T	10893T	131 Gly→Val	(26)
"Cairo"	404 A→C	10905C	135 Asn→Thr	(27)
"Ilesha"	466 G→A	10967A	156 Glu→Lys	(10)
Mahidol	487 G→A	11658A	163 Gly→Ser	(28)
Plymouth	488 G→A	11659A	163 Gly→Asp	(19)
"Chinese-3"	493 A→G	11664G	165 Asn→Asp	(29)
Naone	497 G→A	11668A	166 Arg→His	(17)
"Volendam"	514 C→T	11684T	172 Pro→Ser	(30)
"Nankang"	517 T→C	11687C	173 Phe→Leu	(31)
"Shinshu"	527 A→G	11698G	176 Asp→Gly	(32)
Chikugo	535 A→T	11706T	179 Ser→Cys	(33)
Malaga	542 A→T	11713T	181 Asp→Val	(34)
Santamaria	542 A→T 376 A→G	11713T 10877G	181 Asp→Val 126 Asn→Asp	(35)
Tsukui	561-563 del	11732-11734 del	188 or 189 Ser →del	(22)
Mediterranean Dallas Birmingham "Sassari" "Cagliari" Panama	563 C→T	11734T	188 Ser→Phe	(10) (36) (36) (37) (37) (13)
"Coimbra" "Shunde"	592 C→T	11763T	198 Arg→Cys	(38) (39)
"Santiago"	593 G→C	11764C	198 Arg→Pro	(40)
"Sibari"	634 A→G	11805G	212 Met→Val	(41)
Minnesota Marion Gastonia "LeJeune"	637 G→T	11808T	213 Val→Leu	(42) (42) (42) (43)

Harilaou	648 T→G	11996G	216 Phe→Leu	(44)
"Mexico City"	680 G→A	12028A	227 Arg→Gln	(40)
A- 	680 G→T 376 A→G	12028A 10877G	227 Arg→Leu 126 Asn→Asp	(11)
"Asahikawa"	695 G→A	12043A	232 Cys→Tyr	(33)
Durham	713 A→G	12061G	238 Lys→ Arg	(45)
"Stonybrook"	724-729 GGCACT del	12072-12077	242-243 Gly&Thr del	(46)
Wayne	769 G→C	12117C	257 Arg→Gly	(43)
"Cleveland" Corum	820 G→A	12533A	274 Glu→Lys	(46) (19)
Wexham	833 C→T	12546T	278 Ser→Phe	(19)
"Chinese-1"	835 A→T	12548T	279 Thr→Ser	(47)
Seattle Lodi "Modena" Ferrara II Athens-like	844 G→C	12557C	282 Asp→His	(37) (48) (49) (50) (51)
Osaka	853 C→T	12566T	285 Arg→Cys	(9)
"Montalbano"	854 G→A	12567A	285 Arg→His	(52)
Viangchan Jammu	871 G→A	13031A	291 Val→Met	(43)
"West Virginia"	910 G→T	13070T	303 Val→Phe	(46)
Seoul	916 G→A	13076A	306 Gly→Ser	(33)
Kalyan Kerala	949 G→A	13109A	317 Glu→Lys	(53)
A- Betica Selma Guantanamo	968 T→C 376 A→G	13128C 10877G	323 Leu→Pro 126 Asn→Asp	(11) (14) (14) (54)
"Nara"	953-976 del	13113-13136 del	319-326 del	(55)
Chatham	1003 G→A	13163A	335 Ala→Thr	(10)
"Fushan"	1004 C→A	13164A	335 Ala→Asp	(46)
"Chinese-5"	1024 C→T	13184T	342 Leu→Phe	(26)
Partenope	1052 G→T	13351T	351 Gly→Val	(56)
"Ierapetra"	1057 C→T	13356T	353 Pro→Ser	(40)

Loma Linda	1089 C→A	13388A	363 Asn→Lys	(42)
Calvo Mackenna	1138 A→G	13437G	380 Ile→Val	(57)
Riley	1139 T→C	13438C	380 Ile→Thr	(57)
"Olomouc"	1141 T→C	13440C	381 Phe→Leu	(46)
Tomah	1153 T→C	13452C	385 Cys→Arg	(10)
Iowa Walter Reed Iowa City Springfield	1156 A→G	13455G	386 Lys→Glu	(58)
Guadalajara	1159 C→T	13458T	387 Arg→Cys	(40)
"Mt. Sinai"	1159 C→T 376 A→G	13458T 10877G	387 Arg→Cys 126 Asn→Asp	(59)
Beverly Hills Genova Worcester	1160 G→A	13459A	387 Arg→His	(58) (60) (13)
"Praba"	1166 A→G	13465G	389 Glu→Gly	(46)
Wisconsin	1177 C→G	13476G	393 Arg→Gly	(57)
Nashville Anaheim "Calgary" "Portici"	1178 G→A	13447A	393 Arg→His	(42) (42) (13) (61)
Alhambra	1180 G→C	13479C	394 Val→Leu	(40)
"Bari"	1187 C→T	13486T	396 Pro→Leu	(62)
"Puerto Limon"	1192 G→A	13491A	398 Glu→Lys	(35)
"Anadia"	1193 A→G	13492G	398 Glu→Gly	(63)
Clinic	1215 G→A	13514A	405 Met→Ile	(64)
Riverside	1228 G→T	13527T	410 Gly→Cys	(58)
"Japan" "Shinagawa"	1229 G→A	13529A	410 Gly→Asp	(40) (32)
Tokyo	1246 G→A	13545A	416 Glu→Lys	(19,65)
"Georgia"	1284 C→A	13560A	428 Tyr→End	(46)
"Vansdorf"	3' intron 10 splice site del	13689-13690 del	N/A	(46)
Pawnee	1316 G→C	13719C	439 Arg→Pro	(40)
Telti Kobe	1318 C→T	13721T	440 Leu→Phe	(19) (66)

"Santiago de Cuba"	1339 G→A	13742A	447 Gly→Arg	(10)
"S. Antioco"	1342 A→G	13745G	448 Ser→Gly	(56)
"Cassano"	1347 G→C	13750C	449 Gln→His	(41)
Union Maewo	1360 C→T	13763T	454 Arg→Cys	(47,67) (41,64)
Andalus	1361 G→A	13764A	454 Arg→His	(68)
"Cosenza"	1376 G→C	13884C	459 Arg→Pro	(41)
Canton Taiwan-Hakka Gifu-like Agrigento-like	1376 G→T	13884T	459 Arg→Leu	(69) (70) (69) (69)
Kamiube	1387 C→T	13895T	463 Arg→Cys	(33)
Kaiping Anant Dhon Petrich Sapporo	1388 G→A	13896A	463 Arg→His	(70)
"Fukaya"	1462 G→A	14067A	488 Gly→Ser	(33)
"Campinas"	1463 G→T	14068T	488 Gly→Val	(71)

*According to established convention the names of variants that have not been fully characterized by standardized techniques (72,73) are enclosed in quotes.

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